



## SMOC1 gene

SPARC related modular calcium binding 1

### Normal Function

The *SMOC1* gene provides instructions for making a protein called secreted modular calcium-binding protein 1 (SMOC-1). This protein is found in basement membranes, which are thin, sheet-like structures that support cells in many tissues and help anchor cells to one another during embryonic development. The SMOC-1 protein attaches (binds) to many different proteins and is thought to regulate molecules called growth factors that stimulate the growth and development of tissues throughout the body. These growth factors play important roles in skeletal formation, normal shaping (patterning) of the limbs, as well as eye formation and development. The SMOC-1 protein also likely promotes the maturation (differentiation) of cells that build bones, called osteoblasts.

### Health Conditions Related to Genetic Changes

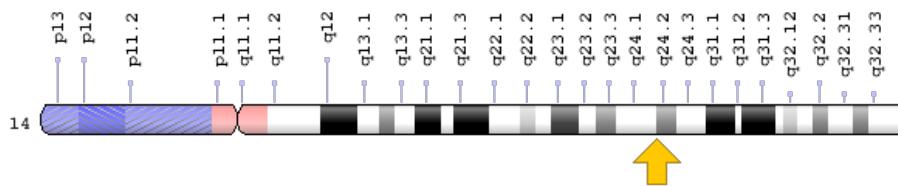
#### ophthalmo-acromelic syndrome

At least 12 mutations in the *SMOC1* gene have been found to cause ophthalmo-acromelic syndrome, a condition that results in malformations of the eyes, hands, and feet. Most of these mutations likely result in a nonfunctional SMOC-1 protein. The loss of SMOC-1 could disrupt growth factor signaling, which would impair the normal development of the skeleton, limbs, and eyes. These changes likely underlie the signs and symptoms of ophthalmo-acromelic syndrome.

## Chromosomal Location

Cytogenetic Location: 14q24.2, which is the long (q) arm of chromosome 14 at position 24.2

Molecular Location: base pairs 69,879,388 to 70,032,366 on chromosome 14 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- secreted modular calcium-binding protein 1
- SMOC1\_HUMAN
- SPARC-related modular calcium-binding protein 1

## Additional Information & Resources

### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28SMOC1%5BTIAB%5D%29+OR+%28SPARC+related+modular+calcium+binding+1%5BTIAB%5D%29%29+OR+%28secreted+modular+calcium-binding+protein+1%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

### OMIM

- SPARC-RELATED MODULAR CALCIUM-BINDING 1  
<http://omim.org/entry/608488>

### Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_SMOC1.html](http://atlasgeneticsoncology.org/Genes/GC_SMOC1.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=SMOC1%5Bgene%5D>

- HGNC Gene Family: SPARC family  
<http://www.genenames.org/cgi-bin/genefamilies/set/1278>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=20318](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=20318)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/64093>
- UniProt  
<http://www.uniprot.org/uniprot/Q9H4F8>

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Reviewed: March 2014  
Published: March 21, 2017

Lister Hill National Center for Biomedical Communications  
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Department of Health & Human Services